SLC5A5 gene

solute carrier family 5 member 5

Normal Function

The *SLC5A5* gene provides instructions for making a protein called sodium (Na)-iodide symporter or NIS. This protein transports iodide, a negatively charged version of iodine, into cells of certain tissues. The NIS protein is found primarily in the thyroid gland, a butterfly-shaped tissue in the lower neck. The thyroid gland produces and releases iodide-containing thyroid hormones that play an important role in regulating growth, brain development, and the rate of chemical reactions in the body (metabolism). The NIS protein supports an efficient system that ensures iodine from the diet accumulates in the thyroid gland for the production of thyroid hormones. This system depends on the NIS protein being positioned in the cell membrane, so it can transport iodide from the bloodstream into particular thyroid cells called follicular cells.

In addition to the thyroid gland, the NIS protein is found in breast tissue during milk production (lactation), ovaries, salivary glands, certain stomach cells (parietal cells), tear glands (lacrimal glands), and a part of the brain called the choroid plexus. During lactation, the NIS protein transports iodide into the milk to supply breast-fed infants with this critical component of thyroid hormones.

Health Conditions Related to Genetic Changes

congenital hypothyroidism

Several *SLC5A5* gene mutations have been identified in people with congenital hypothyroidism, a condition characterized by abnormally low levels of thyroid hormones starting from birth. About half of these mutations delete part of the *SLC5A5* gene or disrupt protein production, resulting in an abnormally small, nonfunctional protein. The remaining mutations change one of the building blocks (amino acids) used to make the NIS protein. Some amino acid substitutions prevent the NIS protein from being positioned in the cell membrane, disabling iodide transport. Other amino acid substitutions do not affect the membrane location of the NIS protein but change the protein's 3-dimensional shape, which impairs its function.

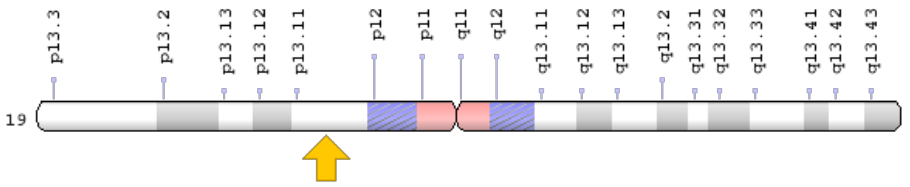
SLC5A5 gene mutations reduce or prevent iodide transport. As a result, the thyroid gland cannot accumulate iodide efficiently, which decreases the production of thyroid hormones. The signs and symptoms of congenital hypothyroidism associated with these gene mutations range from mild to severe depending on the level of hormone production remaining. In many cases, the thyroid gland is enlarged (goiter) in an attempt to compensate for reduced hormone production. Because cases caused by

SLC5A5 gene mutations are due to a disruption of thyroid hormone synthesis, they are classified as thyroid dyshormonogenesis.

Chromosomal Location

Cytogenetic Location: 19p13.11, which is the short (p) arm of chromosome 19 at position 13.11

Molecular Location: base pairs 17,871,394 to 17,895,175 on chromosome 19 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- NIS
- SC5A5_HUMAN
- sodium-iodide symporter
- solute carrier family 5 (sodium/iodide cotransporter), member 5

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28SLC5A5%5BTIAB%5D%29+OR+%28NIS+AND+sodium%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D

OMIM

- SOLUTE CARRIER FAMILY 5 (SODIUM IODIDE SYMPORTER), MEMBER 5
  http://omim.org/entry/601843

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/SLC5A5ID44476ch19p13.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SLC5A5%5Bgene%5D
- HGNC Gene Family: Solute carriers
  http://www.genenames.org/cgi-bin/genefamilies/set/752
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=11040
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/6528
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https://ghr.nlm.nih.gov/gene/SLC5A5

Reviewed: September 2015
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
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